

CASE REPORT

TREACHER COLLINS SYNDROME: A RARE CASE REPORT

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ABSTRACT: PURPOSE: To present a case of Treacher Collins syndrome. **METHODS:** A 7 days-old girl child with eye and facial disfiguration since birth was examined and managed conservatively. **RESULTS:** The presenting symptom of the patient was eye and facial disfiguration .she had sunken appearance of the face, with abnormally wide fish like mouth (macrostomia), depressed nasal bridge, parrot beak nose, malar hypoplasia with an anti mongoloid slant with associated micrognathia, glossoptosis, and high arched palate with cleft palate. On ocular examination, there was notching of lower eyelid, downward slanting eyes, complete absence of lower eyelid lashes. The patient was managed conservatively with nutritional support through nasogastric tube feeding, parenteral antibiotics and eye drops. During discharge, necessary counseling was done with an advice to have a regular follow up for further treatment of hearing loss and reconstructive surgery of facial anomalies. **CONCLUSION:** We are reporting this case of Treacher Collins syndrome baby, with no family history, presenting with the typical orofacial implications of this syndrome.

KEYWORDS: cleft palate, coloboma lid, macrostomia, micrognathia, treacher collins syndrome.

INTRODUCTION: Treacher Collins syndrome is a complex disorder characterized by hypoplasia of facial bones (mandible, maxilla, cheek bones), antimongoloid slant of palpebral fissures, coloboma of the lower lid, deafness, and bilateral anomalies of the auricle. The syndrome was coined after British Ophthalmologist Edward Treacher Collins, who enumerated the features of this syndrome in his paper in 1900. ¹ The incidence of Treacher Collins is only 1 in 50, 000 live births. Diagnosis is made on the basis of clinical signs and symptoms. A treatment protocol based on multi-disciplinary approach can present with complete restoration of the forms and functions of the patient, helping the individual to live a better life.

CLINICAL PRESENTATION: A 7 days old female baby born at 37 week s of gestation via emergency caesarean section, due to pre-eclampsia toxemia to a 23 year old mother, was brought to eye OPD with a complain eye and facial disfiguration since birth. Her family history was unremarkable. The baby cannot close both of her eyes. There was constant watering from both eyes. Gradually the eyes turned dry. Subsequently, there was redness developing in her both eyes. Associated with it the child had breathing and feeding difficulty. The baby had choking and coughing out violently during feeding.



Fig. 1: Patient during first presentation at OPD

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On examination, the patient was euthermic with heart rate 110 beats per minute. Baby was fairly hydrated; she had sunken appearance of the face, with abnormally wide fish like mouth (macrostomia), depressed nasal bridge, parrot beak nose, malar hypoplasia with an anti mongoloid slant. There was associated micrognathia and glossoptosis. There was high arched palate with cleft palate.



Fig. 2: Ocular presentations of the patient

On ocular examination, there was notching of lower eyelid, downward slanting eyes, and complete absence of lower eyelid lashes. There was absence of puncta in lower eyelid. There was epiphora due to atresia of lacrimal duct.



Fig. 3: Otological presentations of the patient

Preauricular skin tag was present in front of both pinna with malformation of both external ears, microtia. The baby was diagnosed as Treacher Collins syndrome based on the clinical features.

The patient was shifted to pediatric department on the same day where she was managed conservatively with nutritional support through nasogastric tube feeding, parenteral antibiotics and moxifloxacin eye drops and carboxy methylcellulose eye ointment. Patient had total oral feeding with satisfactory weight gain with minimal respiratory distress on 12th day of admission. The patient was discharged after 20 days. During discharge, necessary counseling was done with an advice to have a regular follow up for further treatment of hearing loss and reconstructive surgery of facial anomalies.

DISCUSSION: Treacher Collins syndrome, also known as mandibulofacial dystosis, is genetic disorder giving rise to craniofacial malformations. The affected structural development results in problems affecting the form and function of the eyes, ears, nose, maxilla, palate, mandible, and airway.

It has an autosomal dominant inheritance with incomplete penetrance with variable expressibility. An affected parent transmits the disease to 50% of their children according to mendelian laws of genetics. The gene responsible for the disorder is TCOF1, which is located in chromosome 5q31.3 - 5q33.3, which encodes for nucleoprotein named treacle that causes ribosomal

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DNA gene transcription through its interaction with upstream binding factor. This protein during embryogenesis gives rise to facial bones and soft tissues. Mutation in TCOF1 genes reduces the amount of treacle that is produced in the cells and develops the disorder which includes abnormal craniofacial development.² There are no preferences for genders and races.

Patients with Treacher Collins syndrome have a varied Periorbital and ophthalmological presentations. In our case we found down slanting palpebral fissures absent medial lower lid eyelashes, lower lid lacrimal deformity, epiphora, lower lid coloboma. These findings are in accordance to the findings of other investigators.³ Ophthalmological problems included astigmatism, hypermetropia, and squint. Few of the patient consequently developed amblyopia.

Patients with Treacher Collins syndrome may have abnormally small or completely absent external ears. Otological findings potentially associated with TCS include; absent, abnormally small or underdeveloped external ears, atresia or stenosis of the external auditory canals, malformed external ears, Conductive hearing loss attributed to malformations of structures within the middle ear.⁴ Approximately 60% of Patients with Treacher Collins syndrome have a varied dental abnormalities including tooth agenesis, enamel opacities, Widely-spaced teeth, Abnormal eruption of certain teeth (ectopic eruption of maxillary first molars), Malocclusion (improper positioning of the teeth and jaw).⁴ Additional symptoms and physical features may be associated with TCS include; A highly arched roof of the mouth (palate), Cleft palate, Nasal deformity, An abnormally wide mouth (macrostomia), Abnormal scalp hair pattern (front of the ears and extending toward the cheeks), Congenital heart defect.⁴

Radiographs and CT are usually done to evaluate for craniofacial abnormalities. An audiometric evaluation is mandatory as most of the cases develop hearing loss. DNA analysis in most cases (90-95%) cases shows mutation in TCOF1 gene. Prenatal diagnosis during pregnancy can be done by 2 D or 3 D sonology, where presence of polyhydramnios can raise suspicion of the disease or the typical facial morphology can lead to the diagnosis. In cases amniocentesis or CVS can also be a helpful aid in diagnosing the disease.

A multidisciplinary approach mandates for the treatment of the symptoms. Patients with compromised airway patency manifesting as severe respiratory distress since birth, a tracheostomy is performed (and may remain for several years, until the lower jaw has sufficiently grown or until alveolar distraction is performed to enable passage of air through the oral cavity).⁵ Hearing aids of infants are necessary to build up normal bonding process in family and to set up communication skill. Based on the physiological need and development correlating up with the anatomical deformities surgical intervention is planned and executed.

Corrective lenses, contact lenses and other supportive techniques aid in management of Ocular abnormalities. Lubricating Eye ointments may be used, especially at night, to prevent the eyes from drying out during sleep. Corrective reconstructive Surgery may be necessary for correction of a lower lip coloboma. This also helps to improve the facial expression. In case of misaligned eyes where strabismic amblyopia may set to poor vision, strabismic surgery is helpful. Obstructed tear ducts may also require surgery in some cases.⁶

CONCLUSION: Treacher Collins syndrome is an autosomal dominant disorder resulting in abnormalities of structures developing from 1st and 2nd brachial arches. A multidisciplinary approach is required for the patient for functional and psychological development. We hereby report a case of 7 days old baby presenting with the typical orofacial implications of this syndrome.

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